

Attitudes of Genetic Counselors Towards Expanding Newborn Screening and Offering Predictive Genetic Testing to Children

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There is movement to expand newborn screening (NBS) to include conditions that challenge the traditional public health screening criteria. Little is known about the attitudes of genetic counselors towards expanding NBS and offering predictive genetic tests to children. For our study genetic counselors completed an internet survey posted on the National Society of Genetic Counselors Listserv regarding five conditions: cystic fibrosis (CF), Duchenne muscular dystrophy (DMD), glucose-6-phosphate dehydrogenase deficiency (G6PD), fragile X (FraX), and type 1 diabetes (T1D). The survey addressed attitudes towards: (1) testing high-risk infants; (2) mandatory NBS; (3) population screening beyond the newborn period; and (4) testing one's own child. Two hundred sixty-seven usable surveys were received. Over two-thirds of respondents supported testing high-risk infants for all conditions except T1D (22%). CF was the only condition for which there was majority support for both mandatory NBS (56%) and later population screening (60%). For all other conditions, later population screening

was preferred over NBS ($P \leq 0.01$). Genetic counselors were most likely to test their own child for CF (46%) and least likely to test their own child for T1D (6%). For each condition, genetic counselors were more likely to support NBS if they chose to screen their own newborn ($P < 0.001$). Attitudes towards NBS were not influenced by year of graduation or professional experience. We can conclude that genetic counselors are supportive of targeted testing of high-risk infants. They prefer voluntary population screening with consent to mandatory NBS for conditions that challenge Wilson and Jungner criteria. Their support for NBS correlates with their interest in testing their own children and not with professional experience. © 2006 Wiley-Liss, Inc.

Key words: genetic counseling; newborn screening; cystic fibrosis; glucose-6-phosphate dehydrogenase deficiency; fragile X; duchenne muscular dystrophy; type 1 diabetes; attitudes; genetic testing; public policy

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INTRODUCTION

Newborn screening (NBS) is a public health initiative aimed at reducing the morbidity and mortality of congenital conditions [American Academy of Pediatrics (AAP) Committee on Bioethics, 2001; Therrell, 2001; Green and Pass, 2005]. Since its inception with PKU in the 1960s, NBS has expanded and some states are currently screening for over 40 disorders [Green and Pass, 2005]. The completion of the Human Genome Project, the development of high-throughput techniques such as Tandem Mass Spectrometry (MS/MS) and DNA microarrays, and the further identification of disease-causing genes

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and abnormal protein profiles mean that the number of conditions that can be tested for in the newborn period will continue to increase [Cunningham, 2002]. With these opportunities come questions of how to determine what tests should be added to the NBS panel.

Until recently, there was general consensus that NBS be focused on conditions that present in childhood, and for which there is early, efficacious treatment, and clear benefit to the child [Wilson and Jungner, 1968; Institute of Medicine (IOM), 1994; American Academy of Pediatrics (AAP) Newborn Screening Task Force, 2000]. However, NBS programs are now expanding to include conditions that challenge traditional public health criteria as expounded by Wilson and Jungner [Therrell, 2001; Bailey, 2004]. The main force behind this trend is parent advocacy organizations, although they are supported and encouraged by some experts in the field [Howse and Katz, 2000; Natowicz, 2005; Howell, 2006]. The trend creates ethical challenges regarding (1) the role of consent in pediatric genetic testing, particularly for NBS; (2) whether the availability of efficacious treatment is critical to justify NBS; (3) the extent to which an understanding of the natural history and the sensitivity and specificity of the test are necessary for implementing new programs; (4) the psychosocial implications of expanding NBS beyond the traditional paradigm; and (5) the costs and who will incur these costs [Elias and Annas, 1994; Therrell, 2001; Grimes and Schulz, 2002; Ross, 2002; Laberge et al., 2004; Kenner and Moran, 2005].

There are scant data on the attitudes of health care providers towards expanding NBS beyond the traditional criteria [Acharya et al., 2005; Koopmans and Ross, 2006]. To-date, no studies have looked specifically at the attitudes of genetic counselors towards these issues. It is worthwhile to assess their attitudes towards expanding NBS given their unique perspective and their specialized roles within the health care team. Genetic counselors often work directly in NBS programs or in the management of affected children. They are trained in medical genetics, as well as in the psychosocial and ethical issues related to clinical genetics, and serve as educators and advocates for clients and families. They have also taken an active role in creating and promoting ethically responsible policy in the genetics community [National Society of Genetic Counselors (NSGC), 2006].

This study assessed genetic counselors' attitudes towards expanding universal (often mandatory) NBS for five conditions: cystic fibrosis (CF), Duchenne muscular dystrophy (DMD), fragile X syndrome (FraX), type 1 diabetes mellitus (T1D), and glucose-6-phosphate dehydrogenase deficiency (G6PD). It also assessed genetic counselors' attitudes toward testing high-risk children for these conditions, population screening with consent later in infancy,

and testing their own child(ren) in the newborn period. We had four hypotheses. First, genetic counselors would support testing of high-risk infants. Second, genetic counselors' support for expanding screening would correlate with professional experience. Third, genetic counselors would support voluntary population screening for conditions that do not meet Wilson and Jungner criteria, that is, for conditions (1) that have a later age of onset; (2) for which treatment is equivocal or unavailable; or (3) for which the testing may yield ambiguous results. Fourth, genetic counselors' attitudes toward NBS would be influenced by their interest in screening their own children.

METHODS

We administered a questionnaire to genetic counselors using an internet survey (see the online Appendix at <http://www.interscience.wiley.com/jpages/1552-4825/suppmat/index.html>). An e-mail link to the survey was posted by a genetic counselor (K.K.) on the National Society of Genetic Counselors (NSGC) ListServ. The NSGCLIST is an electronic discussion group accessible only by NSGC members. According to the NSGC [2004] Annual Report, there are 2,126 members of the NSGC, and 1,381 genetic counselors are part of the ListServ (personal communication with Jenny Snider, January 2006). SurveyMonkey.com was used to design and administer the survey. The survey (and informed consent agreement) was posted for 2 months (September 13, 2005 to November 16, 2005). Two reminder e-mails with the survey link were posted on the listserv. All responses were anonymous. Genetic counselors were required to consent to the study before completing the survey.

Genetic counselors were queried about five different conditions: CF, G6PD, DMD, FraX, and T1D. These conditions were chosen because they are all being considered for inclusion in various NBS programs and they all have available screening tests, but their potential integration into screening panels challenges the currently accepted boundaries regarding population screening. At the time of our survey, 14 states screened for CF, 2 states screened for G6PD, and 2 states had pilot research projects for T1D screening. No states screened for FraX or DMD, although two pilot research screening programs for DMD were in the planning stages and have now been implemented (GA and OH). Of the conditions included in the survey, only CF was recommended for inclusion in a uniform NBS panel that was commissioned by Health and Research Service Administration [American College of Medical Genetics (ACMG)/Health Resources and Service Administration (HRSA), 2005].

We provided a brief description of each condition, including its inheritance pattern and treatment alternatives. For CF, the genetic counselors were told that nutritional supplementation after early

diagnosis has been shown to improve height and weight, while the pulmonary benefits of early diagnosis are more equivocal. They were also told that the effect of early diagnosis on long-term survival is still unknown. For G6PD, genetic counselors were told that most affected individuals remain asymptomatic until specific triggers are encountered. In addition, they were told that neonatal screening and health education have been shown to be effective in some countries but that screening is rare in the United States because most affected individuals only have a mild form of the disease. For DMD, they were told that some clinicians treat affected patients with steroids to retard the rate of muscular weakness, although others believe that the risks of obesity and decreased bone mineralization outweigh the benefits. For FraX, they were told that early diagnosis can lead to early enrollment in early intervention development programs. For T1D, genetic counselors were told that the presence of certain genetic alleles may predispose or protect individuals from developing the disease. However, it was emphasized that the majority of individuals with high-risk genotypes do not develop the disease and some children with protective genetic alleles do develop the disease. They were also told that preventative treatment is not available and that identification of genotypes is used only for research purposes.

For each condition, genetic counselors were asked about their attitudes towards: (1) testing high-risk infants (infants whose parents were carriers), (2) mandatory universal NBS, and (3) voluntary or mandatory population screening at a later time (beyond the first month of life). For X-linked conditions (G6PD, FraX, and DMD), genetic counselors were given the option of population screening to include only boys or boys and girls. Genetic counselors were also asked about whether they would support screening their own child for each condition. Genetic counselors had the option of making comments after each question. Demographic information was also collected.

Statistical analyses were performed using SPSS14.0. Tables (2 × 2) were analyzed for statistical significance using χ^2 analyses with $P < 0.05$. For all statistical analyses, non-response and the response "Not sure" were excluded. For statistical analysis of these questions, we compared counselors who chose each option, and grouped together all counselors in favor of screening. All additional comments were coded by both S.H. and L.F.R. and then were compared to resolve discrepancies.

This study was approved by the Northwestern University Institutional Review Board and the University of Chicago Institutional Review Board.

RESULTS

Two hundred seventy-five genetic counselors consented to participate, and we received 267 usable

TABLE I. Demographics of Respondents

| Demographic | Characteristic | n (%) |
|---|----------------------------|----------------------|
| Gender (N = 239) | Female | 232 (97) |
| Year of graduation (N = 230) | After 1997 | 155 (67) |
| Specialty* (N = 220) | Prenatal | 134 (50) |
| | Pediatrics | 115 (43) |
| | Adult | 65 (24) |
| | Cancer | 60 (23) |
| | Molecular | 27 (10) |
| | Neurology | 19 (7) |
| | Teratology | 25 (9) |
| | Psychiatric | 1 (0.4) |
| | Infertility | 11 (4) |
| | Involved in NBS: (N = 239) | Previous involvement |
| Current involvement | | 58 (24) |
| No involvement | | 136 (57) |
| Counseled >4 patients with condition* (N = 267) | CF | 125 (47) |
| | G6PD | 32 (12) |
| | FraX | 121 (45) |
| | DMD | 103 (39) |
| | T1D | 45 (17) |
| Family member with this condition (N = 267) | CF | 4 (1.5) |
| | G6PD | 2 (0.7) |
| | FraX | 4 (1.5) |
| | DMD | 4 (1.5) |
| | T1D | 22 (8.2) |

*Total adds up to >100% as respondents were told to mark all that apply.

surveys, providing a response rate of 19.3%. Table I summarizes the demographic information of our respondents. The majority (97%) of respondents were female, and had graduated from their genetic counseling training programs before 1998 (67%). While half described themselves as currently involved in prenatal screening, 39% of respondents had previous or current involvement in NBS programs. One hundred seventy-five counselors (66%) had counseled more than four patients for at least one of these conditions. Twenty-four counselors (9%) reported to have at least one of these diseases in their family; the vast majority having T1D within their families.

Table II describes genetic counselors' attitudes towards different testing scenarios. A general trend was observed across all testing scenarios with decreasing support being expressed when moving from CF to G6PD to FraX to DMD to T1D. In addition, a trend of decreasing support was also observed across all diseases, except for T1D, when moving from testing those at high risk, to population screening, to mandatory NBS, to testing one's own child. Most genetic counselors supported high-risk testing for all conditions except for T1D; this support was particularly pronounced in the case of CF (94%). Later population screening was supported by the majority for CF (60%) and G6PD (54%). CF was the only condition for which over half of the respondents

TABLE II. Genetic Counselors' Attitudes Regarding Testing During Different Time Periods for Each of the Five Conditions

| | Type of testing or screening (n = 267 for all conditions for all questions) | | | | | | | | | | | |
|-------|---|----------|------------------|----------------------------------|----------|------------------|-------------------------|----------|------------------|---------------------------------|----------|------------------|
| | Testing high-risk infants n (%) | | | Screening later in infancy n (%) | | | Newborn screening n (%) | | | Testing your own Children n (%) | | |
| | Yes | No | Not sure/missing | Yes | No | Not sure/missing | Yes | No | Not sure/missing | Yes | No | Not sure/missing |
| CF | 252 (94) | 5 (2) | 10 (4) | 160 (60) | 63 (24) | 44 (16) | 150 (56) | 49 (18) | 68 (26) | 124 (46) | 95 (36) | 48 (18) |
| G6PD* | 231 (87) | 6 (2) | 30 (11) | 144 (54) | 64 (24) | 59 (22) | 105 (39) | 82 (31) | 80 (30) | 52 (20) | 171 (64) | 44 (17) |
| FraX | 196 (73) | 29 (11) | 42 (16) | 96 (36) | 120 (45) | 51 (19) | 53 (20) | 147 (55) | 67 (25) | 39 (15) | 179 (67) | 49 (18) |
| DMD* | 182 (68) | 45 (17) | 40 (15) | 68 (25) | 151 (57) | 48 (18) | 38 (14) | 174 (65) | 55 (21) | 20 (8) | 190 (71) | 57 (21) |
| T1D* | 54 (20) | 152 (57) | 61 (23) | 62 (23) | 153 (57) | 52 (20) | 15 (6) | 208 (78) | 44 (17) | 17 (6) | 210 (79) | 40 (15) |

*Total may not add up to 100% due to rounding.

TABLE III. Support for Screening Your Own Child Versus Support for NBS

| Do you support screening your own child? n (%) | Do you support NBS? n (%) | |
|--|---------------------------|----------|
| | Yes | No |
| CF (n = 173) | | |
| Yes | 100 (90) | 11 (10) |
| No | 30 (48) | 32 (52) |
| G6PD (n = 166) | | |
| Yes | 41 (95) | 2 (5) |
| No | 50 (41) | 73 (59) |
| FraX (n = 182) | | |
| Yes | 21 (72) | 8 (28) |
| No | 24 (16) | 129 (84) |
| DMD (n = 186) | | |
| Yes | 7 (41) | 10 (59) |
| No | 21 (12) | 148 (88) |
| T1D (n = 215) | | |
| Yes | 7 (50) | 7 (50) |
| No | 6 (3) | 195 (97) |

$\chi^2, P < 0.001.$

(56%) supported mandatory NBS. In all scenarios except T1D, least support was expressed for testing one's own child. For T1D, there was equal lack of support for mandatory NBS and testing one's own child (6%). CF received the most support for testing one's own child (46%).

For each condition, there was a significant association between support for screening one's own child and support for NBS ($P < 0.001$) (Table III). Similarly, those who did *not* support screening their own child were also more likely to not support NBS for all of the conditions. Support for NBS for each condition was not significantly associated with previous or current involvement in NBS, year of graduation, or having counseled more than four clients for this condition. Counselors who were not working in pediatrics were more likely to support NBS for DMD ($P \leq 0.025$) but not for any other condition ($P = NS$); (data not shown).

Table IV displays support for testing (high risk, population screening, and NBS) for the X-linked conditions G6PD, FraX, and DMD. For those who support testing for these conditions, the majority advocated G6PD and FraX testing for both boys and girls in all testing scenarios. For DMD, respondents were more evenly divided in their support for testing both boys and girls versus testing only boys. There was almost equal support for testing all children versus testing only boys in the case of NBS (53% vs. 47%, $P = NS$), and exactly equal support in the case of later population screening.

Table V describes those who support later population screening. The majority of these respondents support voluntary screening for all conditions, with support ranging from 87 to 94%. Preference for voluntary or mandatory population screening was

TABLE IV. Testing Boys Versus Boys and Girls for X-Linked Conditions in High-Risk Infants (HR), Mandatory Newborn Screening (NBS), and Population Screening Later (POP)

| Do you support testing for? n (%) | G6PD | | | FraX | | | DMD | | |
|-----------------------------------|--------------|---------------|---------------|--------------|--------------|--------------|--------------|--------------|--------------|
| | HR (n = 231) | POP (n = 143) | NBS (n = 105) | HR (n = 196) | POP (n = 96) | NBS (n = 53) | HR (n = 183) | POP (n = 68) | NBS (n = 38) |
| Boys and girls | 150 (65) | 111 (78) | 79 (75) | 168 (86) | 86 (90) | 50 (94) | 63 (35) | 34 (50) | 20 (53) |
| Only boys | 81 (35) | 32 (22) | 26 (25) | 28 (14) | 10 (10) | 3 (6) | 119 (65) | 34 (50) | 18 (47) |

not significantly associated with working in pediatrics, year of graduation from genetic counseling programs, or with experience counseling for that particular condition (data not shown).

Almost half of the respondents (127, 47.6%) made a total of 589 comments. Comments were well-distributed over the five conditions, with most frequent comments about CF (164 or 22.8%) and least frequent regarding T1D (97 or 15.6%). The themes were varied focusing on the specifics of each condition as well as more global problems with counseling and testing. The CF comments largely focused on the nature of the testing; the methodology (DNA-based vs. biochemical), the sensitivity and specificity of the test, and the false positive and false negative rates. Concerns pertaining to sensitivity and specificity of testing were also common in references to G6PD. For the DMD questions, most comments addressed the equivocal nature of treatment. FraX comments also focused on the lack of clear medical benefit available from early diagnosis. DMD and FraX questions were also most likely to elicit comments pertaining to reproductive matters, including reproductive planning (DMD) and carrier screening for women in the prenatal period (FraX). Comments for T1D questions focused on the poor predictive value of testing, the lack of confirmed genotype–phenotype correlations, and the use of testing as part of a research protocol. In addition, psychosocial issues of parental confusion, anxiety, and false reassurance were common. Comments regarding targeted testing of high-risk infants and at-risk ethnicities were most often cited in reference to CF and G6PD. There were also 133 comments directed towards issues of consent. The majority of these (84, 63%) were made in reference to promoting voluntary consent in high-risk testing.

TABLE V. Of Those who Support Population Screening, Preference for Voluntary Versus Mandatory

| | Voluntary n (%) | Mandatory n (%) |
|----------------|-----------------|-----------------|
| CF (n = 160) | 142 (89) | 18 (11) |
| G6PD (n = 144) | 125 (87) | 19 (13) |
| FraX (n = 96)* | 85 (89) | 11 (12) |
| DMD (n = 68) | 63 (93) | 5 (7) |
| T1D (n = 62)* | 58 (94) | 4 (7) |

*Total may be >100% due to rounding.

DISCUSSION

These are the first data collected regarding the attitudes of genetic counselors to testing and screening newborns for a variety of pediatric health conditions. The majority of genetic counselors who participated in our study preferred targeting testing to high-risk individuals (based on family history, ethnic group, or symptomatology) rather than universal population screening for the conditions addressed in the survey. Targeted testing provides the advantage of increasing the sensitivity of the test. In addition, children with positive family histories may already be viewed as at-risk by their parents. Focusing on the high-risk community could minimize the potential negative effects of providing an increased risk result to an unsuspecting family [Ross, 2003]. Targeting rather than universal screening may be preferable for the conditions included in the survey because they do not fulfill all of the Wilson and Jungner criteria for population screening.

The wide support for targeted testing based on risk factors, however, was limited to conditions that would provide a diagnosis (CF, G6PD, FraX, DMD), and was not expressed for pre-dispositional testing (T1D). Lack of support for pre-disposition testing for T1D may be related to the nature of the test, the lack of available preventative and treatment options, and the potential harms of pre-disposition testing, which include increased anxiety, misunderstanding of test results, interference with parent–child bonding, stigma, and cost [Wertz et al., 1994; American Society of Human Genetics (ASHG)/American College of Medical Genetics (ACMG), 1995; Clayton, 1999; Ross, 2002].

CF was the only condition for which there was majority support for both mandatory NBS (56%) and population screening at a later date (60%). Later population screening was preferred to NBS for all other conditions ($P \leq 0.01$). The benefits of later population screening include greater opportunity for education and pre- and post-test counseling. The temporal separation also helps distinguish these less traditional tests from state-mandated screening programs [Acharya et al., 2005]. However, there are additional costs to later population screening including the need for a second blood sample and a separate infrastructure from the current NBS programs [Acharya et al., 2005].

The greater support for population screening later in infancy rather than in the newborn period in our study may be related to a difference in the way these testing options were presented in our survey. Participants were told that NBS was provided as a universal mandatory program, whereas they were given the option of choosing voluntary or mandatory testing in the case of later population screening. Of those who supported population screening, the large majority (>87% for each condition) advocated voluntary rather than mandatory screening. Thus, greater support for later population screening may actually reflect a preference for voluntary screening rather than preference in timing. This would be consistent with the general ethos of genetic counseling which advocates a process of informed decision-making, with an emphasis on the preservation of patient autonomy [Walker, 1998; Veach et al., 2003]. Genetic counselors may believe that the process of informed consent is particularly critical for those conditions that do not clearly meet the traditional criteria for public health screening.

For all conditions except T1D, the genetic counselors expressed a stronger preference for population screening (in the newborn period or later) than testing one's own child. These findings are consistent with data reporting that health care practitioners are likely to grant patients' requests for testing and treatment options that they would not necessarily choose for themselves or their children [Gonen et al., 2002; Campbell and Ross, 2003; Acharya et al., 2005]. This could reflect a health care provider's respect for patient autonomy by supporting the patient or parent's right to choose options that may differ from their own personal desires [Gonen et al., 2002]. Alternatively, willingness to accede to patient requests may also signify concerns about the legal implications of not supporting patient decisions [Wilfond and Thomson, 2000].

Genetic counselors' support for screening both boys and girls versus screening only boys varied among the X-linked conditions. Genetic counselors expressed support for screening both genders for FraX, a finding which was also observed among pediatricians [Acharya et al., 2005] and may reflect acknowledgment that females can be affected by the condition [Crawford et al., 2001; McConkie-Rosell et al., 2005] and can benefit from early intervention [Bailey, 2004]. Genetic counselors' support for screening both boys and girls for G6PD may again be due to the fact that both boys and girls can develop symptoms of jaundice in the newborn period or have a hemolytic crisis secondary to exposures [Kaplan and Hammerman, 2000]. In contrast, there is limited medical benefit of identifying female DMD carriers in childhood because manifesting carriers are quite rare [Hoogerwaard et al., 1999]. And yet, about half of the genetic counselors who supported DMD screening would include females, which was also similar to

pediatricians' attitudes [Acharya et al., 2005]. As mentioned by some respondents, the support may be attributed, in part, to the potential reproductive benefit of identifying carrier mothers. Identifying carrier females could also offer these girls reproductive knowledge that will be useful to them in adulthood. These comments are in opposition to the consensus positions that state that genetic testing of children for their own reproductive benefit should be delayed until they can make their own informed decision [Institute of Medicine (IOM), 1994; American Society of Human Genetics/American College of Medical Genetics, 1995; American Academy of Pediatrics (AAP) Committee on Bioethics, 2001; Canadian Paediatric Society (CPS), 2003].

We found a significant association between genetic counselors' support for NBS and their desire to screen their own child for each condition ($P < 0.001$ for all conditions). Pediatricians have also been found to be more likely to support NBS for a condition if they advocated testing their own child for that condition [Acharya et al., 2005]. The correlation between genetic counselors' support for testing their own child and support for implementing universal NBS suggests that genetic counselors' personal preferences may color their professional policy recommendations. This finding is not completely unexpected in light of the growing recognition of how genetic counselors' values and opinions influence their counseling style and the decisions of their patients [Anderson, 1999; Rentmeester, 2001], but emphasizes the need for evidence in policy making rather than policies being based mainly on professional recommendations [Natowicz, 2005; Botkin et al., 2006]. And yet, to-date, most NBS programs have been expanded under an extemporaneous model based on stakeholder advocacy rather than using an evidentiary model where policies are based on epidemiological and clinical data [Wilfond and Thomson, 2000, Natowicz, 2005]. At minimum, our data support Botkin's argument that a methodological evaluation of screening tests and programs are necessary [Botkin, 2005].

Although the correlation between support for screening one's own child and support for NBS holds for each condition, the degree of support varied greatly across conditions. For those who would screen their own child for CF or G6PD, a large majority (90 and 95%, respectively) supported NBS. In contrast, those who would screen their own child for DMD or T1D expressed weaker support for NBS (41 and 50%, respectively).

The stronger support for CF and G6PD NBS could reflect the belief that the benefits of screening are great enough as to warrant universal mandatory screening for the general population. Even if counselors chose not to screen their own children for CF or G6PD, they still recognized some utility to population screening. As in the case of CF, almost

half of the genetic counselors who did not want to screen their own child still supported universal NBS for CF. One possible explanation for this finding is that genetic counselors may not feel the need to screen their own children for CF because they have plans to screen themselves prenatally. Alternatively, concerns about justice may lead them to support NBS for CF because they recognize that many women do not seek prenatal care and do not have access to prenatal testing [Koopmans and Ross, 2006].

The weaker support for DMD and T1D NBS could be explained by those who saw benefit in screening their own child for these conditions, but recognized that this benefit was not sufficient as to warrant mandatory population screening. Those who did not want to screen their own children for these conditions strongly rejected NBS for DMD (88%) and T1D (97%), perhaps reflecting an awareness of the significant potential harms to this testing. They expressed concern that there is no clinical benefit to early diagnosis of DMD and T1D, and that most individuals at risk for T1D never develop the disease.

For FraX, those who supported screening their own child strongly supported NBS (72%), and those who did not want to screen their own children were strongly opposed to NBS (84%). These responses represent two very distinct views on NBS for FraX, and the complex risks and benefits associated with this screening [Bailey, 2004; McConkie-Rosell et al., 2005]. Those who support NBS see potential for early intervention; those who oppose fear the risks of labeling and self-fulfilling prophecies [Cohen, 1998; Ross, 2002].

Our survey was limited by its recruitment through the NSGC listserv which only yielded a 19% response rate. Of course, it is not known how many subscribers are actually active subscribers. Despite this, our demographics are reflective of the genetic counselors who responded to the 2004 NSGC Professional Status Survey (2004) supporting the external validity of our findings. A second limitation was the multiple choice design of our survey which potentially could have constrained responses and masked nuances. However, we did provide the opportunity for respondents to make comments for most of the questions, and we did incorporate this qualitative data into our results and discussion.

CONCLUSION

Genetic counselors are supportive of targeted testing of high-risk infants. They prefer voluntary population screening with consent to mandatory NBS for conditions that challenge Wilson and Jungner criteria. Their support for NBS correlates with their interest in testing their own children and not with professional experience. This affirms the

potential for bias of policies based on professionals' opinions rather than evidence.

This survey is only a preliminary step. Further study is needed to understand the influence of the personal on professional guidelines and practice, as well as to explore the similarities and differences between health care professionals from different specialties who offer genetic testing and screening.

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